Interaction between hereditary spherocytosis and the beta-thalassemia trait: A case report

Kalıtsal sferositoz ve beta-talasemi taşıyıcılığı arasındaki etkileşim: Olgu sunumu

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To the Editor,

Coinheritance of hereditary spherocytosis (HS) and β thalassemia is very rare. HS is a familial haemolytic disorder resulting from primary abnormality of red cell membrane. It is transmitted as an autosomal dominant trait. β thalassemia is also a common inherited disorder. In Indians, the frequency of β thalassemia is reported between 3.5&14.9% [1]. The haemolytic anemia resulting from their coexistence has been shown to be of variable severity in different studies [2-5].

We hereby present a case of a 50 yr old Nepalese female who came with fever, cough & weakness without any organomegaly. CBC findings revealed microcytic hypochromic red cells with high red cell count ($>5x10^6/\mu l$) and mildly increased RDW suggestive of βTT (Table 1). Peripheral smear showed large number of microspherocytes, microcytic hypochromic cells, target cells and occasional red cells with basophilic stippling (Figure 1). Reticulocyte count was 1.5%. Direct Coomb's test was negative and serum bilirubin was normal (1.2 g/dl). HPLC of Hb revealed an increased Hb A2 (4.8%) and Hb F (6.0%). Her son revealed very few spherocytes in peripheral smear and CBC findings were suggestive of βTT. Coomb's test was negative, Hb HPLC showed high HbA2 (5.1%). The incubated osmotic fragility curves of both the patient and her son were shifted to right with a tail of fragile cells. Thus, a diagnosis of HS with βTT was made in both (Figure 2).

Inheritance of HS has been reported in association with α thalassemia, β thalassemia and certain enzyme deficiencies [2-8].

The results are conflicting regarding the degree of hemolysis, when hereditary spherocytosis and

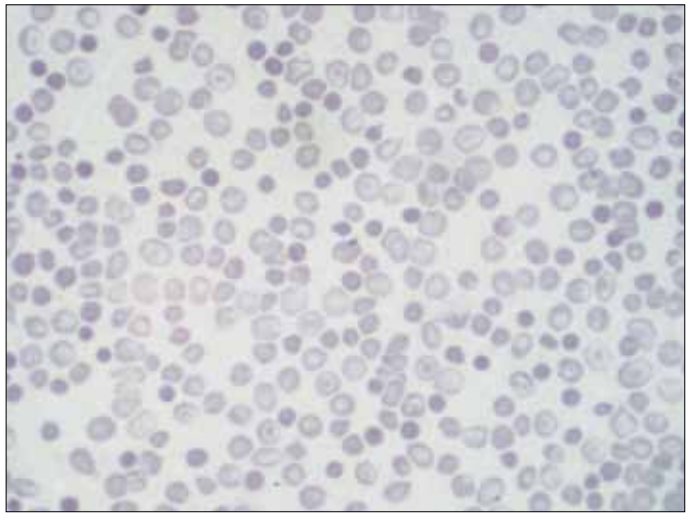

Figure 1. (400X)-Peripheral smear (Wright's stain) showing large number of microspherocytes and some target cells

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Table 1. Hematological parameters

Parameter	Patient	Son	Husband
Hb g/dl	10.3	12.1	14.0
RBC count x 10^{12}/l	5.37	6.26	5.05
Hct %	34.5	39.9	43.0
MCV (fl)	64.2	63.7	85.1
MCH (pg)	19.2	19.3	28.0
MCHC (g/dl)	29.9	30.3	32.9
RDW (%)	20.7	16.1	14.4
TLC /cumm	8,500	4,800	9,000
Plt Count x10^9/l	149	142	154
Hb HPLC			
HbA%	89.2	94	98.9
HbA2%	4.8	5.1	1.5
HbF%	6.0	0.9	0.4

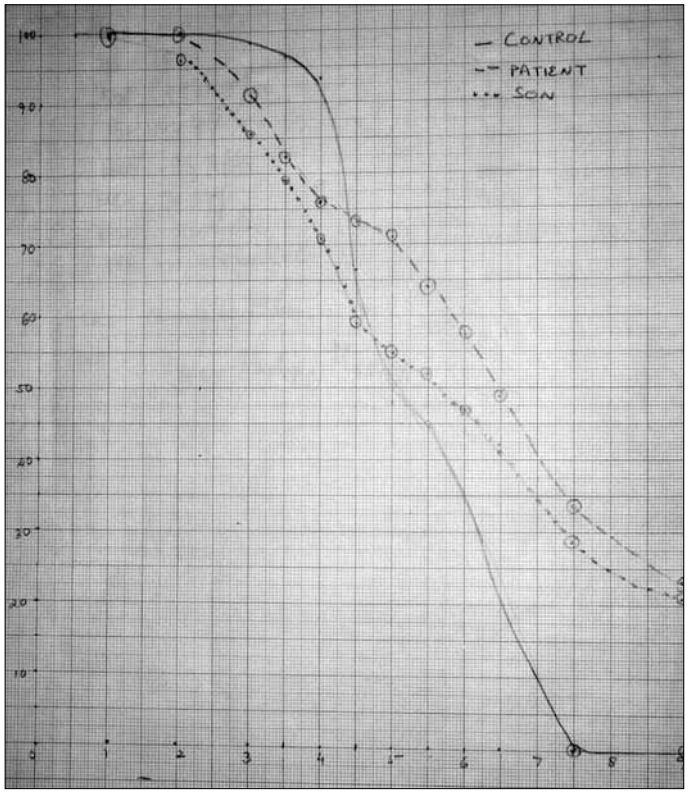

Figure 2. Incubated Osmotic fragility curve showing shift to the right compared to the control

heterozygous β thalassemia coexist. In our case the peripheral smear showed a large number of microspherocytes, pointing towards moderate HS (20-30/hpf). Moderate HS is associated with a chronic haemolytic anemia with modest splenomegaly and intermittent jaundice. However, our patient was asymptomatic till date. This corollary can be explained by assuming that the coinheritance of βTT with HS probably had an influence on clinical outcome. The microcytic hypochromic red cells of βTT and spherocytes of HS had opposite properties with regards to their fragility and this probably leads to reduced severity of hemolysis. Hence, if both HS and βTT coexist, the later silences the HS and ameliorates the degree of hemolysis.

Written informed consent was obtained from the patient.

Conflict of interest statement

The authors of this paper have no conflicts of interest, including specific financial interests, relationships, and/or affiliations relevant to the subject matter or materials included.

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